Letter to Editor

Peutz–Jeghers Syndrome

Dear Editor,

PeutzJeghers Syndrome (PJS) is an inherited autosomal dominant disorder, and attributed to a mutation localized at 19p13.3.[1] This condition is characterized by mucocutaneous pigmentation and gastrointestinal polyps. The purpose of reporting this case is to remind clinicians that perioral or hand pigmentation should not be taken lightly in colorectal cases [Figure 1]. PJS also assumes more importance because of higher risk of intestinal and extraintestinal cancers.[2] Histologically, these lesions have an arborzing pattern with fronds covered by normal epithelium that incorporates all the cell types located at the site of origin. Endocrine cells and smooth muscle form an important component of the lesions.

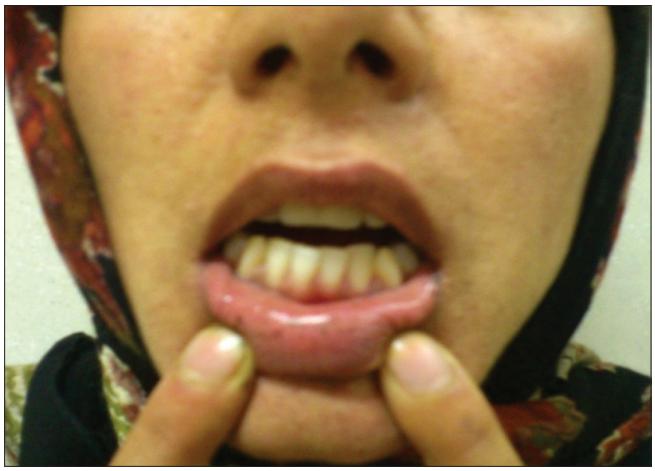

Figure 1: Clinical photograph showing peri-oral mucosal pigmentation

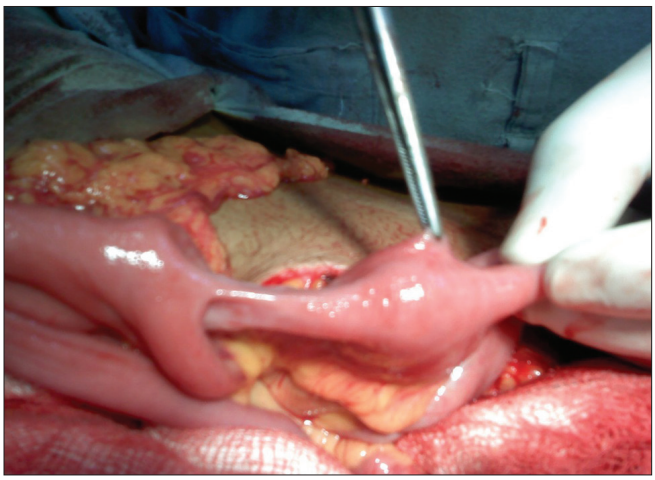

Figure 2: Operative photograph showing the lead point of intussusception in small gut

A 26 year-old female presented to us with history of bleeding per rectum and a mass coming out per rectum for last 2 years. Colonoscopy revealed a small polypoidal lesion at 5 cm and an extensive polypoidal growth at 10–18 cm from anal verge. Upper gastrointestinal endoscopy revealed a 3-cm polyp in the antrum and a 1.5-cm sessile polyp in the second part of duodenum. Antral polyp was resected endoscopically. Computerized tomogram enterography revealed more polyps in the upper small gut. The operative findings revealed three more polyps in the small gut, with one of them causing intussusception. A few enlarged perirectal and mesenteric nodes were also found. A small polyp and a big polypoidal mass were present in mid rectum. Intussusception was reduced and multiple enterotomies were performed to excise small gut polyps. Low anterior resection was done for excising the polypoidal mass and the rectal polyp [Figure 2]. Histopathology revealed morphology and histology of Peutz–Jeghers polyp, showing a core of broad bands of muscularis mucosa smooth muscle fibers, the hallmark of a PJS polyp with no evidence of malignancy.

The relationship of mucocutaneous pigmentation and intestinal polyposis was first reported in 1921 by Peutz,[3] who studied a family over three generations. Peutz noticed that seven of them had intestinal polyposis and four had nasal polyposis, besides the peculiar mucocutaneous pigmentation. After an initial report of two patients by Jeghers in 1944,[4] the definitive clinical description of PJS was written by Jeghers *et al*. in detail in 1949.[5] The authors also recognized that it was inherited as a simple Mendelian dominant trait. The majority of patients meeting the clinical diagnostic criteria have a causative mutation in the *STK11* gene,[6] which is located at 19p13.3.[1] The cancer risks in this condition are substantial, particularly for breast and gastrointestinal cancer.[7] In view of an established risk of cancers in PJS patients, many centers subject the patients to surveillance using capsule endoscopy. However, a prospective study[8] revealed that MR enterography offers a promising alternative to capsule endoscopy for small bowel polyp surveillance in adults with PJS. Laparoscopy is a minimally invasive approach that deals with the acute problem and may prevent adhesion formation and subsequent repeat laparotomy for small bowel obstruction. Our patient is on follow-up from last 8 months and is doing well.

Fazl Qadir Parray, Abdul Wahid Syed, Ghulam Nabi Yatoo[1], Showkat Ali Zargar[1], Rayees Ahmad Malik[2]

Department of Surgery, [1]Department of Medical GE, [2]Department of Pathology, Sher-i-Kashmir Institute of Medical Sciences, Soura, Srinagar, J and K, India.
E-mail: fazlparray@rediffmail.com

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